



SHH gene

sonic hedgehog

Normal Function

The *SHH* gene provides instructions for making a protein called Sonic Hedgehog. This protein functions as a chemical signal that is essential for embryonic development. Sonic Hedgehog plays a role in cell growth, cell specialization, and the normal shaping (patterning) of the body. This protein is important for development of the brain and spinal cord (central nervous system), eyes, limbs, and many other parts of the body.

Sonic Hedgehog is necessary for the development of the front part of the brain (forebrain). This signaling protein helps establish the line that separates the right and left sides of the forebrain (the midline). Specifically, Sonic Hedgehog establishes the midline for the underside (ventral surface) of the forebrain. Sonic Hedgehog and other signaling proteins are needed to form the right and left halves (hemispheres) of the brain.

Sonic Hedgehog also has an important role in the formation of the eyes. During early development, the cells that develop into the eyes form a single structure called the eye field. This structure is located in the center of the developing face. Sonic hedgehog signaling causes the eye field to separate into two distinct eyes.

Health Conditions Related to Genetic Changes

coloboma

microphthalmia

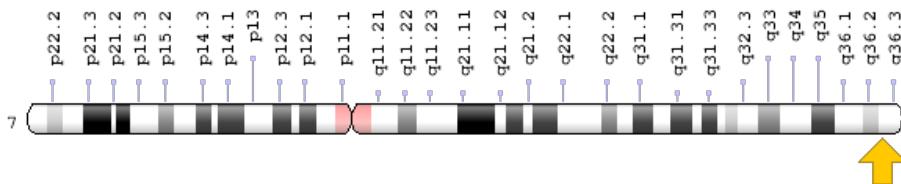
nonsyndromic holoprosencephaly

More than 100 mutations in the *SHH* gene have been found to cause nonsyndromic holoprosencephaly. This condition occurs when the brain fails to divide into two hemispheres during early development. *SHH* gene mutations are the most common cause of nonsyndromic holoprosencephaly. These mutations reduce or eliminate the activity of Sonic Hedgehog. Without the correct activity of this protein, the eyes will not form normally and the brain does not separate into two hemispheres. The development of other parts of the face is affected if the eyes do not move to their proper position. The signs and symptoms of nonsyndromic holoprosencephaly are caused by abnormal development of the brain and face.

Chromosomal Location

Cytogenetic Location: 7q36.3, which is the long (q) arm of chromosome 7 at position 36.3

Molecular Location: base pairs 155,799,984 to 155,812,273 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- HPE3
- SHH_HUMAN
- sonic hedgehog homolog (Drosophila)
- sonic hedgehog protein
- sonic hedgehog protein preproprotein

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): The Hedgehog Pathway
<https://www.ncbi.nlm.nih.gov/books/NBK10043/#A1063>

GeneReviews

- Holoprosencephaly Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1530>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SHH%5BTI%5D%29+OR+%28sonic+hedgehog%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- SONIC HEDGEHOG
<http://omim.org/entry/600725>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/SHHID378.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SHH%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10848
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6469>
- UniProt
<http://www.uniprot.org/uniprot/Q15465>

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